# Appendix

# DHEC Newborn Screening Contact Information

## Bureau of Maternal and Child Health (MCH), Children's Health and Perinatal Services

**Please call the staff listed below if you have any questions about newborn screening clinical or follow-up services:**

Medical Consultant: Eileen Walsh, MD.....803-898-0362
email: walshem@dhec.sc.gov

Newborn Screening (NBS) Follow-up Director:
Tanya Spells, MS, MT(ASCP).....803-898-0619
email: spellsty@dhec.sc.gov

NBS Follow-up Program Coordinator, Hospital QI contact, Abnormal Results:
Dana Smith, RN, BSN.....803-898-0593
email: smithdm@dhec.sc.gov

NBS Follow-up Program Nutritionist, Metabolic Formula, Abnormal Results:
Jennifer Schlub, RD, LDN……..803-898-1969
email: schlubjk@dhec.sc.gov

NBS Follow-up Program Coordinator, Abnormal Results:
Stephanie Bachman, MT(ASCP).....803-898-3845
email: bachmasj@dhec.sc.gov

NBS Administrative Assistant (Temp):
Karla Clyde…….803-898-3192
Email: clydeka@dhec.sc.gov

Fax number for all Children's Health/Newborn Screening staff.....803-898-0337

Mailing address for all Newborn Screening Follow-up Program staff:

(NAME)
SC DHEC Children's Health
Newborn Screening Program
2100 Bull Street
Columbia, SC 29201

## Public Health Laboratory (PHL)

**Please call the staff listed below if you have any questions about newborn screening laboratory services:**

Newborn Screening Patient Lab Results:

Request NBS Patient Lab Results (fax)……803-896-0298
Lab Information Management Systems (LIMS fax)….803-896-3862

Testing/Technical questions:

Chemistry Division Director
Elizabeth Bair……803-896-0991
Email: bairea@dhec.sc.gov

Newborn Screening Lab Manager
Sandi Hall, MT(ASCP)…….....803-896-0891
email: hallss@dhec.sc.gov

NBS Lab Supervisor
Linda Conway, MLT(ASCP)……803-896-5938
Email: conwayll@dhec.sc.gov

NBS Lab Supervisor
Graham McCaskill……803-896-0878
Email: mccasksg@dhec.sc.gov

NBS Lab QI Technologist
Amanda Jenkins, MT(ASCP)...……..803-896-0935
Email: jenkinja@dhec.sc.gov

Fax number for all Public Health Laboratory staff.....803-896-0983

Mailing address for Newborn Screening Public Health Laboratory staff:

(NAME)
SC DHEC PHL
Chemistry Division
Newborn Screening Laboratory
8231 Parklane Road
Columbia, SC 29223

QA and On-site Training Workshops:

Quality Manager
Patricia Myers, MT(ASCP).....803-896-3897
Email: myerspa@dhec.sc.gov

Lynn Gleaton, MT(ASCP)……...803-896-0899
Email: gleatoll@dhec.sc.gov

Laboratory Information Management Systems (LIMS):

LIMS Administrator
Shatoya Wright, MBA…….....803-896-4777
Email: wrightsl@dhec.sc.gov

Melonie Carter, MS……...803-896-0945
Email: carterma@dhec.sc.gov

Fax number for LIMS Department……..803-896-3862

To order Newborn Screening Collection Forms and Mailing Envelopes:

Lab Supply Manager
Marcus Hudson.....803-896-0913
email: hudsonmm@dhec.sc.gov

Data Entry and Lab Billing:

Support Manager
Melissa Dawson.....803-896-2331
email: dawsonmf@dhec.sc.gov

## Educational Materials Library (EML) and Forms

Contact:

Lamar O'Neil….....803-898-3539
Fax for all ERC staff……...803-898-3476

Website: http://www.scdhec.gov/Agency/EML/

To order NBS brochures and forms:

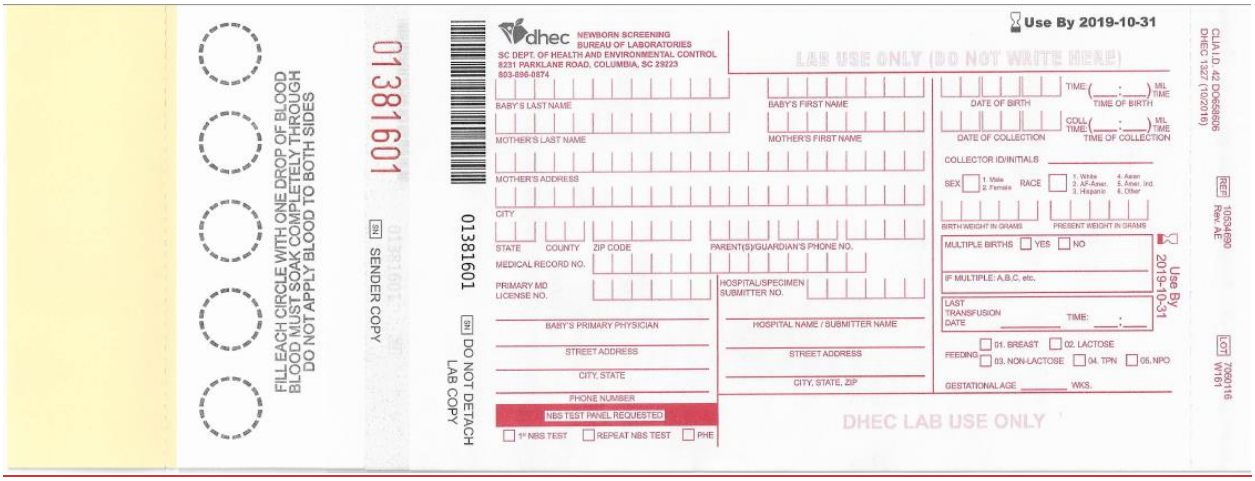
Newborn Screening Brochures "*For Your Baby's Health*" (ML-000032 in English)
Newborn Screening Brochures "*Para la salud de su bebe*" (ML-025096 in Spanish)

Parental Statement of Religious Objection Form (DHEC 1804 in English)

Parental Statement of Religious Objection Form "*Declaracion de objection religiosa por parte de los padres*" (DHEC 1804S in Spanish)

Mailing address for Education Materials Library staff:

SC DHEC/EML
2600 Bull Street
Columbia, SC 29201




SOUTH CAROLINA DEPARTMENT OF HEALTH AND ENVIRONMENTAL CONTROL
PUBLIC HEALTH LABORATORY

Newborn Screening (Instructions for Completing DHEC-1327) Revised 10/2016

**PURPOSE**

Newborn Screening on newborns in South Carolina is a State Law. This form is used to provide identification and essential information and a means of submitting blood samples for newborn screening. Due to the makeup of this form and the information needed, it cannot be pre-addressed by the Public Health Laboratory. It must be filled out completely by the provider.

NOTE: There is a space for two senders. Both senders will receive a copy of the results.

A completed form must be submitted with the circles on the filter paper filled with the newborn's blood. The instructions for specimen collection and handling of blood specimen are on the back of the form.

Never place the form in plastic bags to submit to the laboratory. Plastic bags can cause false laboratory results. Always check expiration date of the filter paper. This information is on the face of the form. **The laboratory will not accept blood on expired filter paper forms**.

Follow the general instructions for the patient and sender information. Further instructions are below.

BABY'S LAST NAME: Enter baby's legal last name

BABY'S FIRST NAME: Enter baby's legal first name

MOTHER'S LAST NAME: Enter mother's last name, adoption agency or lawyer's office (if considered baby's legal guardian).

MOTHER'S FIRST NAME: Enter mother's first name.

MOTHER'S ADDRESS: Enter mother's complete mailing address, city, state, county code, and zip code. (See back of the sender copy for county codes)

PARENT(S)/GUARDIAN'S PHONE NO.: Enter telephone number of parent(s) or guardian.

HOSPITAL DHEC NO.: Enter hospital medical record number or DHEC Patient Number.

PRIMARY MD LICENSE NO. (BABY'S PRIMARY PHYSICIAN): Enter the number assigned by the SC State Board of Medical Examiners of South Carolina preceded by the letter "M". If in a group of physicians enter the number assigned by the Public Health Laboratory preceded by the letter "G".

BABY'S PRIMARY PHYSICIAN: Enter doctor's name, Street Address, City, State, and phone number.

BILLING NUMBER: Used by Health Department.

PROGRAM NUMBER: Used by Health Department

HOSPITAL/SPECIMEN SUBMITTER NO.: Enter the sender number. If a hospital, enter the number assigned by the Public Health Laboratory preceded by the letter "H".

HOSPITAL NAME/SUBMITTER NAME.: Enter name of hospital or medical group/MD that is submitting the specimen.

STREET ADDRESS: Enter hospital or medical group/MD street address.
CITY, STATE, ZIP: Enter hospital's or medical group/MD's city, state and zip code.

NBS TEST PANEL REQUESTED: Check whether it is the 1st NBS TEST PANEL, REPEAT NBS TEST PANEL or PKU (PHE) sample only.

**NOTE: PKU is only one of the 50+ NBS screening tests available. Marking "PKU" when ALL tests are needed will mean a complete screening may NOT be performed**.

DATE OF BIRTH: Enter baby's date of birth. Enter month, day, and year. Precede all numbers less than ten (10) with a zero (0). Example: September 1, 2017 would be 09/01/17.

TIME: Enter time of birth (hour and minute). USE MILITARY (24 HOUR CLOCK) TIME. Precede all numbers less than ten (10) with a zero (0). Examples: 9:20 am would be 09:20. 9:20 pm would be 21:20.

SEX: Enter "1" for Male or "2" for Female in the block.

RACE: Insert appropriate number in block as outlined below:

| | |
|---|---|
| 1. White | 4. Asian |
| 2. African-American | 5. American Indian |
| 3. Hispanic | 6. Other |

BIRTH WEIGHT IN GRAMS: Enter weight of baby at birth in GRAMS.

PRESENT WEIGHT IN GRAMS: Enter weight of baby at time of specimen collection in GRAMS.

MULTIPLE BIRTHS: Mark an "X" in the appropriate box Yes or No.

IF MULTIPLE: A, B, C, etc.: If multiple birth – YES, write in baby's birth order (i.e., A, B, C, etc.)

LAST TRANSFUSION DATE: If baby has received any blood product containing red cells (including in utero transfusions), write in the date of the last transfusion (month, day, and year).

FEEDING: Check the appropriate box

DATE COLLECTED: Enter month, day, and year specimen was collected. Precede all numbers less than ten (10) with a zero (0). Example: September 1, 2017 would be 09/01/17.

TIME OF COLLECTION: Enter time of collection (hour and minute). USE MILITARY (24 HOUR CLOCK) TIME. Precede all numbers less than ten (10) with a zero (0). Examples: 9:20 am would be 09:20. 9:20 pm would be 21:20.

THE FORM: The form is made up of three parts:

Part 1: Lab copy. DO NOT detach.
Part 2: Sender's copy can be retained by the sender.
Part 3: The Cover, along with Part 1, must stay attached and be returned to the laboratory. The flap over the dried blood spots must cover the spots when the NBS form is placed in the envelope for mailing.

DO NOT USE TAPE or STAPLES on the form. DO NOT USE an addressograph on the form. The addressograph can compress the filter paper and mar the blood.

DO NOT write or place labels in the top area of the form that is designated "LAB USE ONLY."

OFFICE MECHANICS AND FILING: After processing in the laboratory, a computer-generated report will be mailed to the sender(s) and the laboratory will retain the original paperwork.

## Best Specimen Collection Timing by Disorder

| Condition | Best Age to Screen | Factors Affecting Tests | Consequences to Infant if Not Identified |
|---|---|---|---|
| Biotinidase deficiency | Birth - 72 hours | False positive-premature/jaundiced<br><br>False negative-red cell transfusion/ECLS (extracorporeal life support) | Hypotonia, seizures, developmental delay, abnormal movements, breathing problems, hair loss and hearing loss |
| Congenital Adrenal Hyperplasia | 12-48 hours and 2-4 weeks | False positive-sick/stressed infant<br><br>False negative-maternal steroids; infant dexamethasone | Acute crisis with failure to thrive, dehydration and shock, early puberty, virilization of females<br><br>Can cause death in the newborn period |
| Cystic Fibrosis | 24 hours - 7 days | False positive-hypoxia, respiratory stress, hypoglycemia, trisomies (13, 18, 21), preterm, collect <12 hours<br><br>False negative-meconium ileus; other GI | Failure to thrive, malnutrition, severe respiratory disease |
| Congenital Hypothyroidism | 12-72 hours and 2-6 weeks | False positive-sick/stressed infant, preterm, topical iodine<br><br>False negative-delayed TSH rise | Prolonged jaundice, lethargy, poor muscle tone, Intellectual disability (ID), abnormal movements, motor delays |
| Fatty Acid Oxidation Disorders | Birth - 48 hours | False positive-Carnitine or MCT supplementation<br><br>False negative-Carnitine supplementation | Hypoketotic hypoglycemia, metabolic decompensation/crisis, seizures<br><br>Can cause death in the newborn period |
| Galactosemia | Birth - 48 hours | False positive-liver disease<br><br>False negative-red cell transfusion/ECLS extracorporeal life support | Hypoglycemia, jaundice, sepsis, failure to thrive, Intellectual disability (ID)<br><br>Can cause death in the newborn period |

| Condition | Best Age to Screen | Factors Affecting Tests | Consequences to Infant if Not Identified |
|---|---|---|---|
| Hemoglobinopathies | Birth - 72 hours | False positive-none reported<br><br>False negative-red cell transfusion/ECLS (extracorporeal life support) | Chronic hemolysis, intermittent vaso-occlusive pain episodes, splenic dysfunction which can lead to life-threatening infection |
| Urea Cycle, Amino Acid, and Organic Acid Disorders | 24-48 hours | False positive-PN, liver disease, immature liver enzymes<br><br>False negative-collect <24 hours | Seizures, lethargy, poor feeding, metabolic decompensation/crisis, coma, developmental delay, Intellectual disability (ID)<br><br>Some forms can cause death in the newborn period |

**Adapted from *Newborn Screening for Preterm, Low Birth Weight, and Sick Newborns; Approved Guideline,* Clinical and Laboratory Standards Institute, October 2009**

## Criteria for Notification of Abnormal Results: Weekday/Monday Holiday

The following table outlines the methods by which the physician of record is notified by the Division of Children's Health and/or the Public Health Laboratory of an abnormal screening result or an unacceptable specimen, as of 1/5/2019:

| **Condition** | **CRITICAL results: Phone call with fax and mailed confirmation** | **Abnormal results: Mailed confirmation only** |
|---|---|---|
| | | |
| | | |
| CH | TSH >= 40 µIU/mL – *CRITICAL*<br><br>TSH >= 100 µIU/mL - *PANIC* | TSH between 19 and 39 µIU/ml (for infants <= 7 days old) and<br><br>TSH > 10 uIU/ml (for infants > 7 days old) |
| Galactosemia | GALT < 3.00 U/dL, regardless of GAO result | GAO >= 12.0 mg/dL with normal GALT |
| | | GALT from 3.00 to 3.75 U/dL, regardless of GAO result |
| CAH | 17-OHP >= 48 ng/mL for infants with birth weights >= 2500 grams<br><br>17-OHP >= 75 ng/mL *CRITICAL* | 17-OHP from 30 to 47 ng/mL for infants with birth weights >= 2500 grams |
| | 17-OHP >= 130 g/mL for infants with birth weights < 2500 grams | 17-OHP from 76 to 129 g/mL for infants with birth weights < 2500 grams |
| **Amino Acid disorders** | | |
| PKU | PHE >= 300 | PHE >= 130 |
| TYR I | Elevated SUAC, regardless of TYR result | |
| TYR I or TYR II/III | TYR >= 800 µM regardless of SUAC result | TYR from 330 to 800 µM with normal SUAC result |
| HCY | MET >= 65 | |
| Cbl C, D | MET <= 6 | |
| MSUD | LEU+ILE >= 316 and/or<br>VAL >= 300 | |
| CIT I/II, ASA | CIT >= 100 | CIT >= 60 |
| **Fatty acid disorders** | Most abnormalities (likelihood of disease status in some abnormal acyl carnitines can be determined using the CLIR validated tools) | Results from selected abnormal acyl carnitines that score likely normal using the CLIR validated tools |

| **Condition** | **CRITICAL results: Phone call with fax and mailed confirmation** | **Abnormal results: Mailed confirmation only** |
|---|---|---|
| | | |
| | | |
| CUD | C0 <= 5.00 | C0 <= 8.00 |
| MCAD | C8 >= 0.52<br>C8 >= 1.5 *CRTICAL* | |
| VLCAD | C14:1 >= 0.50 with elevated C14:1/C2 | |
| VLCAD | C14:1 >= 0.75 regardless of C14:1/C2 level<br><br>C14:1 >= 1.5 *CRITICA*L | |
| LCHAD/TFP | C16-OH >= 0.08 | |
| **Organic acid disorders** | Acyl carnitines other than C3: Most abnormalities (likelihood of disease status in some abnormal Acyl carnitines can be determined using the CLIR validated tools) | Results from selected abnormal acyl carnitines that score likely normal using the CLIR validated tools |
| PROP, MUT, MCD, Cbl A, B | None | C3 >= 5.30 |
| MMA, PROP | C3 >= 10 μM with elevated C3/C2 | C3 from 5.30 to 10 μM with elevated C3/C2 |
| PROP, MUT, MCD, Cbl A, B | C3 >= 15 μM with normal C3/C2 *CRITICAL* | C3 from 5.30 to 15 μM with normal C3/C2 |
| *IBG* | | C4 >= 1.30 |
| 3-MCC, HMG, βKT | C4DC+C5OH >= 1.00 *CRITICAL* | |
| GA-I | C5DC+C6OH >= 1.00 *CRITICAL* | |
| IVA, 2-MBG | C5 >= 0.72<br>C5 >= 2.5 *CRITICAL* | |
| GA II | C4 >= 1.30 and C5 >= 0.72 with Multiple elevated acyl carnitines | |
| Cystic Fibrosis - IRT | None | Any elevated IRT |
| Cystic Fibrosis – 2nd tier testing* | 1 or more mutations found* | No mutations found |
| Biotinidase Deficiency | Any abnormal result | |
| Sickling Hemoglobinopathies | Any sickling disease result: FS, FSA, FSB, FSC, FSV, FSE/O or FSD/G<br>(*start Penicillin*) | Email notification to Regional Program Coordinators in Children & Youth with Special |

| Condition | CRITICAL results: Phone call with fax and mailed confirmation | Abnormal results: Mailed confirmation only |
|---|---|---|
| | | |
| | | |
| | | Healthcare Needs (CYSHCN) Sickle Cell Program |
| Non-sickling Hemoglobinopathies | FC, FD, FE/O, FF | |
| Hemoglobin traits and carriers* (ex. FAS) | None | Lab notification to PCP and Regional Sickle Cell Foundations (CBO's) for follow up, education and counseling. |
| Hemoglobin Bart's (FAB) | FA +Bart's >= 15%<br>FA +Bart's >= 25% - *CRITICAL* | FA +Bart's < 15% |
| SCID | TREC > 34 Cq and/or<br>RNase P > 27.5 Cq | |
| Unacceptable specimen | NICU babies only | Parent and Provider notification for any situation where the initial specimen was unacceptable, unless it's a NICU baby. |

***Note: Carrier Status**

Everyone has two copies of each gene, one from their mom and one from their dad. Being a carrier means that one copy of the gene mutated (changed) and was passed on to a person, but the other copy is normal and functioning fine. Since carriers still have one working copy of the gene, they typically do not have health problems associated with carrying a mutation.

Therefore, many people are carriers of a disease-causing mutation without even knowing it. Being a carrier, however, means there is an increased chance that a child could be born with a genetic disease. Anyone can be a carrier of a genetic disease, even if no one in the family is affected.

**Critical and Panic Values**

The SC Newborn Screening Program defines critical values (also known as panic values) as laboratory test results that exceed established limit(s) (high or low) as defined by the laboratory for certain analytes as listed in the Critical (Panic) Limits. Critical results are considered life-threatening and require immediate notification of the physician, the physician's representative, the ordering entity, or other clinical personnel responsible for the patient's care.
Note: Abnormal results are not considered critical values. Results that are outside the laboratory's established reference intervals may be considered abnormal, but the terms "abnormal" and "critical" should not be used interchangeably.

## Criteria for Notification of Abnormal Results: Saturday/Other Holiday

The following table outlines the methods by which the physician of record is notified by the Division of Children's Health and/or the Public Health Laboratory when the result is indicative of immediate morbidity/mortality.

| **Abnormal Analyte(s)** | **Condition(s)** | **Action** |
|---|---|---|
| PHE | PKU | Wait until Monday |
| LEU+ILE and/or VAL | MSUD | Contact MD |
| MET | HCY | Wait until Monday |
| CIT | Citrullinemia I, II, ASA | Contact MD |
| SUAC | TYR I | Contact MD |
| TYR | TYR II, III | Wait until Monday |
| GALT <= 3.00 and high GAO >= 12.0 | Classical Galactosemia | Contact MD |
| GALT <= 1.875 and normal GAO (< 12.0) | Duarte Galactosemia | Contact MD |
| GALT normal (>= 3.75) and GAO >= 24.0 | Other Galactosemia | Contact MD |
| Repeat GALT normal and high GAO >= 12.0 | Other Galactosemia | Contact MD |
| Low C0 | CUD | Contact MD if C0 < 5.00 and C3+C16 < 2 |
| High C0 | CPT IA | Contact MD if indicated by CLIR tools |
| High C0/(C16+C18) | CPT IA | Contact MD if<br>Ratio >= 45 and infant <= 7 days old |
| C3 >10 or C3 >15 with one or more high ratios | PROP, MMA | Contact MD |
| C3 high with both ratios high | PROP, MMA | Contact MD |
| C3 high not detailed above | PROP, Cbl A, B; MCD, MUT | Wait until Monday |
| C3DC+C4OH | MAL | Wait until Monday |
| C4 | *IBG* | Wait until Monday |
| C5 | IVA, 2-MBG | Contact MD |
| C4 and C5 | GA II | Contact MD<br>Use CLIR dual scatter plot tool to differentiate MCAD from GA II |
| Multiple short and medium chain AC's<br>C4, C5 (C8, C10, C12, C14, C14:1, C16OH) | GA II | Contact MD if indicated:<br><br>Use CLIR dual scatter plot tool to differentiate MCAD from GA II |

| Abnormal Analyte(s) | Condition(s) | Action |
|---|---|---|
| C4DC+C5OH | 3-MCC, HMG, MCD, βKT, 3MGA, 2M3HBA | Contact MD if C4DC+C5OH > 1.0 |
| C5DC+C6OH | GA I | Contact MD if C5DC+C6OH >1.0 |
| C8 (C6, C10) | MCAD | Contact MD |
| C10:2 | Dienoyl reductase (DE RED) | Wait until Monday |
| C14:1 | VLCAD | Contact MD if C14:1>=0.75 in isolation or C14:1 >=0.50 with elevated ratio.<br><br>Use CLIR dual scatter plot tool to differentiate VLCAD vs. VLCAD (het) |
| High C16OH and/or C18 | LCHAD, TFP | Contact MD, if indicated by CLIR tool |
| High C16 and C18:1 | CPT II | Contact MD |
| Biotinidase < 70.0 U/dL | Biotinidase deficiency | Wait until Monday |
| Biotinidase < 35.0 U/dL | Biotinidase deficiency | Contact MD |
| TSH < 39 | CH | Wait until Monday |
| TSH >= 40 | CH | Contact MD |
| 17OHP >= 48 in NBW or 130 in LBW | CAH | Contact MD |
| 17-OHP lower level abnormal result | CAH | Wait until Monday |
| Critical elevation of IRT | CF | Contact MD |
| Any CF mutation found | CF or CF carrier | Wait until Monday |
| Abnormal Hgb | Sickling Hgb Disease | Contact MD |
| Abnormal Hgb trait | FA +Bart's >= 25% | Contact MD |
| Abnormal Hgb | Non-sickling Hgb disease | Wait until Monday |
| TREC >= 34 Cq and/or RNase P >= 27.5 Cq | SCID or other Immune disorder | Contact MD |

# Referral sources for Metabolic Disorders in South Carolina

Greenwood Genetic Center—Metabolic Hotline: 866-262-3070

**Dr. Richard Schroer, Sr. Clinical/Biochemical Geneticist**
Greenwood Genetic Center-Charleston
3520 W Montague Ave, Ste 103
N Charleston, SC 29418
Office Number: 877-679-0927
Fax: 866-676-9881
Alternate Fax: 843-735-5095
Pager: 864-942-1315
Cell: 843-206-9037
Geographic areas covered: Low Country/Midlands/Pee Dee

**Dr. Neena Champaigne, Clinical Geneticist**
Greenwood Genetic Center-Greenwood
101 Gregor Mendel Circle
Greenwood, SC 29646
Office Number: 864-388-1064
Toll Free: 888-442-4363
Fax: 864-388-1720
Geographic areas covered: Upstate

**Biochemical Laboratory Resources:**
**Tim Wood, PhD**
Greenwood Genetic Center
Biochemical Genetics Laboratory
106 Gregor Mendel Circle
Greenwood SC 29646
Phone - 864-941-8177
Fax - 864-941-8133
Statewide coverage

Note: This list is not inclusive of all biomedical/genetic specialists in South Carolina.

# Pediatric Referral Sources for Cystic Fibrosis in South Carolina

**Sylvia E Szentpetery MD, MPH**
MUSC Comprehensive Cystic Fibrosis Center
Department of Pediatrics
135 Rutledge Ave, Ste 279
Charleston, SC 29425
Office: 843-876-1555
Fax: 843-876-1583
szentpet@musc.edu

Kimberly Brown Foil
Genetic Counselor
brownkl@musc.edu
843-792-6474

For referrals use MEDULINE: 800-922-5250

**Daniel Brown, MD**
Pediatric Pulmonary Associates
9 Medical Park, Ste 505
Columbia, SC 29203
Office: 803-434-2165
Fax: 803-434-2083
dan.brown@uscmed.sc.edu

**Steven Snodgrass, MD**
Pediatric Pulmonology
200 Patewood Drive, Ste A300
Greenville, SC 29615
Office: 864-454-5533
Fax: 864-241-9246
ssnodgrass@ghs.org

Contact Rebecca Peace - CF coordinator. Her direct number is 864-454-5566.

To schedule a sweat test, call PRISMA Health Greenville (GHS) Lab, Manual chemistry dept. Sweat tests are scheduled M/W/F in Children's Cancer Center

NOTE: This list is not inclusive of all pediatric pulmonologists in South Carolina.

# Referral Sources for Pediatric Hemoglobin Disorders in SC

Greenville Children's Hospital
Pediatric Hematology and Oncology
BI-LO Charities Children's Cancer Center
900 W. Faris Road, 2nd Floor
Greenville, SC 29605
(864) 455-8898

Greenville Children's Hospital
Pediatric Hematology and Oncology
BI-LO Charities Children's Cancer Center
249 N. Grove Medical Park Drive, Suite 200
Spartanburg, SC 29303
(864) 573-8732

Greenville Children's Hospital
Pediatric Hematology and Oncology
BI-LO Charities Children's Cancer Center
2000 E. Greenville Street, Ste 3500
Anderson, SC 29261
(864) 716-6490

http://www.ghschildrens.org/specialists/pediatric-hematology-oncology

Medical University of South Carolina (MUSC) Children's Hospital
Rutledge Tower
135 Rutledge Avenue
Charleston, SC 29425
(843) 876-0444

Medical University of South Carolina (MUSC) Children's Hospital
165 Ashley Avenue
Charleston, SC 29425
(843) 792-1414

https://www.musckids.org/our-services/sickle-cell-center

Palmetto Health Children's Hospital
Center for Cancer and Blood Disorders
7 Richland Medical Park
Columbia, SC 29203
(803) 434-3533
(803) 296-5437
https://www.palmettohealth.org

# Referral Sources for Pediatric Endocrine Disorders in SC

**MUSC Children's Hospital**
Pediatric Endocrinology
135 Rutledge Ave
Charleston, SC 29425
For referrals use MEDULINE: 800-922-5250
Phone: 843-792-6807
Fax: 843-792-0548
Deborah Bowlby, MD

**GHS Children's Hospital**
Pediatric Endocrinology
200 Patewood Drive, Ste 200
Greenville, SC 29615
Phone: 864-454-5100
Fax: 864-241-9238
Elaine M. Apperson, MD

NOTE: This list is not inclusive of all pediatric endocrinologists in South Carolina.

# Referral Sources for Immune Disorders in South Carolina

Lower State: Charleston, Berkeley, Dorchester, Colleton, Hampton, Jasper, Beaufort, Georgetown, Williamsburg, Horry, Marion, Dillon, Marlboro, Darlington, Florence

**Kelli Williams, MD MPH**
Summey Medical Pavilion
2250 Mall Drive
North Charleston, SC 29406
Office: 843-876-0444
Fax: 843-876-1583
For Urgent needs:
MUSC Paging Operator: 843-792-2300

**Maria Streck, MD**
Ben Sawyer Primary Care
1440 Ben Sawyer Blvd, Ste 1109
Mt Pleasant, SC 29464
Office: 843-876-8333
Fax: 843-876-8330

Upstate: York, Chester, Union, Laurens, Greenwood, McCormick, Abbeville, Anderson, Oconee, Pickens, Greenville, Spartanburg, Cherokee

**Joshua Brownlee, MD**
Pediatric Infectious Diseases
GHS Children's Hospital
200 Patewood Dr. Ste A200
Greenville, SC 29615
Office: 864-454-5130
FAX: 864-454-5698

All Counties
**Michelle Hudspeth, MD**
Amanda Little, RN
MUSC Blood and Bone Marrow Transplant Team
MUSC Rutledge Tower
135 Rutledge Ave
Charleston, SC 29425
Office: 843-792-0381
Fax: 843-792-8912

Midstate: Clarendon, Chesterfield, Lee, Sumter, Calhoun, Orangeburg, Bamberg, Allendale, Barnwell, Aiken, Lexington, Richland, Kershaw, Lancaster, Fairfield, Newberry, Saluda, Edgefield, Aiken

**Greg Black, MD**
Carolina Allergy and Asthma Consultants
One Richland Medical Park, Ste 200
Columbia, SC 29203
Office: 803-929-0290
Fax: 803-779-0344

# Newborn Screening Law and Regulation

## Law

Neonatal Screening for Inborn Metabolic Errors and Hemoglobinopathies Section 44-37-30, of the South Carolina Code of Laws

(A) A child born in this State, except a child born of a parent who objects on religious grounds and indicates this objection before testing on a form promulgated in regulation by the Department of Health and Environmental Control, shall have neonatal testing to detect inborn metabolic errors and hemoglobinopathies.

(B) Information obtained as a result of the tests conducted pursuant to this section is confidential and may be released only to a parent or legal guardian of the child, the child's physician, and the child when eighteen years of age or older when requested on a form promulgated in regulation by the department.

(C) A blood sample obtained pursuant to this section is confidential and may be released only as the parent or legal guardian of the child from whom a blood sample was obtained, or the child when eighteen years of age or older, directs the department at the time of testing or at any time after that on a form promulgated in regulation by the department.

(D)(1) Unless otherwise directed pursuant to this subsection, a blood sample obtained pursuant to this section must be stored by the department at minus 20º centigrade and may be released for purposes of confidential, anonymous scientific study. The release of a blood sample must conform with regulations promulgated by the department.

At the time of testing or at any time after that, on a form promulgated in regulation by the department, the parent or legal guardian of the child from whom a blood sample was obtained, or the child when eighteen years of age or older, may direct the department to:

(a) return a blood sample in its entirety and any test results not less than two years after the date of testing;
(b) destroy a blood sample in a scientifically acceptable manner not less than two years after the date of the testing; or
(c) store a blood sample at minus 20º centigrade but not release the blood sample for confidential, anonymous scientific study.

(D)(2) A blood sample released for confidential, anonymous study pursuant to this section must not contain information which may be used to determine the identity of the donor. A blood sample released pursuant to this section may contain demographic or other statistical information.

If scientific study identifies genetic information that may benefit the child, the department may notify confidentially the parent or legal guardian, or the child if eighteen years of age or older, of this information.

(E)(1) A blood sample that has not been stored at minus 20° Centigrade before the effective date of this section must be destroyed in a scientifically acceptable manner six months from the effective date of this section unless a parent or legal guardian of a child from whom a blood sample was obtained, or the child if eighteen years of age or older, requests return of the blood sample on a form provided by the department.

(E)(2) A blood sample stored at minus 20° centigrade pursuant to this section before the effective date of this section must be retained as prescribed in subsection (D) unless directed by the parent or legal guardian of the child from whom a blood sample was obtained to destroy or return the blood sample.

(F) The department shall promulgate regulations necessary for the implementation of this section. All forms must include information concerning the benefits of neonatal testing and storage of a blood sample.

(G) A person who violates this section or the regulations promulgated pursuant to this section or who provides or obtains or otherwise tampers with a blood sample collected pursuant to this section is guilty of a misdemeanor and, upon conviction, may be fined not more than fifty thousand dollars or imprisoned for not more than three years."

Severability

SECTION 3. If any section, subsection, paragraph, subparagraph, sentence, clause, phrase, or word of this act is for any reason held to be unconstitutional or invalid, such holding shall not affect the constitutionality or the validity of the remaining portions of this act, the General Assembly hereby declaring that it would have passed this act, and each and every section, subsection, paragraph, subparagraph, sentence, clause, phrase, and word thereof, irrespective of the fact that any one or more other sections, paragraphs, subparagraphs, sentences, clauses, phrases, or words thereof may be declared to be unconstitutional, invalid, or otherwise ineffective.

Proviso 34.37. (DHEC: Metabolic Screening) The department may suspend any activity related to blood sample storage as outlined in Section 44-37-30 (D) and (E) of the 1976 Code, if there are insufficient state funds to support the storage requirements. In that event, the samples may be destroyed in a scientifically appropriate manner after testing. The department shall notify providers of the suspension within thirty days of its effective date.

# Regulation

South Carolina Department of Health and Environmental Control
REGULATION 61-80
Neonatal Screening For Inborn
Metabolic Errors and Hemoglobinopathies

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**Section A - Purpose and Scope**

This regulation establishes rules implementing provisions of Section 44-37-30 of the South Carolina Code of Laws, 1976, as amended, regarding testing of newborn children for inborn metabolic errors and hemoglobinopathies.

The Department of Health and Environmental Control has been given the legislative mandate to promulgate rules and regulations for screening for inborn metabolic errors and hemoglobinopathies and to ensure compliance with the screening of every child born in South Carolina.

The responsibilities of the various agencies, institutions and persons involved in the screening process are defined. Procedures for storage and use of blood specimens and maintenance of confidentiality are included.

**Section B - Definitions**

1. Inborn Metabolic Errors--shall mean inborn errors of metabolism.

2. Hemoglobinopathy--shall mean a hematologic disorder or carrier state caused by alteration in the genetically determined molecular structure of hemoglobin which may result in overt anemia as well as clinical and other laboratory abnormalities.

3. Identifying Information--shall mean child's legal name, sex, race, birth date, time of birth, place of birth, birth weight, current weight, feeding type; parent's or legal guardian's complete name, complete address and telephone number; mother's Social Security Number.

4. Attending Physician--shall mean the physician who has entered into an agreement to provide care during and/or after delivery for the mother and/or her child. The physician listed on the laboratory form will be assumed to be the attending physician until notification to the contrary is received in accordance with Official Departmental Instructions.

5. Department—shall mean the South Carolina Department of Health and Environmental Control.

6. Laboratory--shall mean the South Carolina Department of Health and Environmental Control Public Health Laboratory.

7. Bureau of Maternal and Child Health--shall mean an organizational unit of the South Carolina Department of Health and Environmental Control.

8. Official Departmental Instructions--shall mean detailed instructions approved by the Commissioner of the South Carolina Department of Health and Environmental Control or his designee under which the public and private health care providers, including hospitals, laboratories, clinics, physicians and their staffs screen all children born in South Carolina for designated Inborn Metabolic Errors and Hemoglobinopathies.

**Section C - Testing**

1. The Laboratory shall perform all screening tests for inborn metabolic errors and hemoglobinopathies using procedures compliant with the Clinical Laboratories Improvement Act (CLIA) of 1988, as amended, and approved by the Food and Drug Administration. If any result is abnormal, the appropriate test shall be repeated and confirmatory tests performed in accordance with Official Departmental Instructions.

2. The Laboratory, in conjunction with the Bureau of Maternal and Child Health, shall adopt standards for the quality assurance and interpretation of approved tests and for the collection of specimens.

3. Confirmation and repeat specimen testing are available from the Laboratory at no charge to patients suspected or diagnosed as having one of the diseases if the analysis is completed at the Laboratory.

4. Test results and identifying information are to be reported and recorded in accordance with Official Departmental Instructions.

**Section D - Collection of Specimen**

1. A specimen shall be collected from every child born in South Carolina for the purpose of screening for inborn metabolic errors and hemoglobinopathies.

2. **Births in a Hospital**

a. The attending physician is responsible for the collection of the specimen from every child born in the hospital in accordance with Official Departmental Instructions and is responsible for submission of the specimen to the Laboratory on the day of collection.

b. Under the direction of the attending physician, the specimen shall be collected under the most favorable conditions following the procedures specified in the Official Departmental Instructions. The brochure produced by the Department that explains newborn screening for inborn metabolic errors and hemoglobinopathies and blood specimen storage options shall be given to the parent or legal guardian of the child.

c. A specimen shall be collected from every child born in the hospital prior to release from the hospital (except when the parents object due to religious convictions) in accordance with the procedure specified in the Official Departmental Instructions. If the parent objects to the screening on the basis of religious convictions, the parent shall complete the procedure specified in the Official Departmental Instructions.

d. If for some reason the specimen is not collected at the hospital, the hospital shall then be responsible for notifying the Bureau of Maternal and Child Health as specified in the Official Departmental Instructions.

e. The Hospital shall review the patient record for each child born in the hospital no later than ten (10) days after delivery to ensure that a specimen was collected and submitted to the Laboratory.

3. **Births Outside a Hospital**

a. The attending physician is responsible for the collection of the specimen from every child in accordance with the Official Departmental Instructions and for submission of the specimen to the Laboratory on the day of collection.

b. Under the direction of the attending physician, the specimen shall be collected under the most favorable conditions following the procedure specified in the Official Departmental Instructions.

The brochure produced by the Department that explains newborn screening for inborn metabolic errors and hemoglobinopathies and blood specimen storage options shall be given to the parent or legal guardian of the child.

c. If the parents object to the screening on the basis of religious convictions, the parents shall complete the procedure specified in the Official Departmental Instructions.

d. If for some reason the specimen is not collected within three (3) days of delivery by the attending physician, this physician shall notify the Bureau of Maternal and Child Health as specified in the Official Departmental Instructions.

e. If there is not an attending physician, then the person in attendance is responsible for the collection of the specimen. If there is no other person in attendance, then the parents or legal guardian shall notify the Health Department in the county in which the child resides within three (3) days of delivery so that a specimen may be collected.

**Section E - Assurance of Diagnosis and Follow-up**

1. Information obtained as a result of the tests conducted for screening for inborn metabolic errors and hemoglobinopathies is confidential and may be released only to the infant's physician or other staff acting under the direction of the physician, the child's parent or legal guardian, and the child when he/she is eighteen years of age or older.

2. Normal and abnormal test results will be forwarded by the Laboratory and/or Bureau of Maternal and Child Health to the attending physician who shall be responsible for informing the parents or legal guardian of test results.

3. If the child is not under the care of the attending physician, as specified in the Official Departmental Instructions, the person in attendance shall notify the Bureau of Maternal and Child Health. The Department will then notify the parents or legal guardian of the test results.

4. Upon notification that a specimen was insufficient or that it is necessary for a test to be repeated, the attending physician shall collect and submit a second specimen to the Laboratory in accordance with Official Departmental Instructions.

5. The attending physician shall initiate appropriate medical follow-up and diagnosis when abnormal test results occur. If that is not possible, the Bureau of Maternal and Child Health shall be notified as specified in the Official Departmental Instructions.

6. The attending physician shall notify the Bureau of Maternal and Child Health of all children born in South Carolina who are diagnosed as having inborn metabolic errors or hemoglobinopathies.

7. Appropriate genetic counseling should be offered to all families of children with abnormal test results as outlined in the Official Departmental Instructions.

**Section F - Storage of Specimens**

1. Hospital staff or other persons who collect blood specimens for the purpose of screening for inborn metabolic errors and hemoglobinopathies shall inform each child's parent or legal guardian of the blood specimen storage options.

2. Hospital staff or other persons who collect these blood specimens shall give the brochure produced by the Department that explains newborn screening for inborn metabolic errors and hemoglobinopathies to the parent or legal guardian as a means of informing them of the benefits of screening and blood specimen storage. Hospital staff or other persons who collect these blood specimens shall indicate that the brochure was given to the parent or legal guardian by documenting in the appropriate space on the Blood Sample Storage Options Form.

3. The Laboratory shall store all specimens at minus 20° Centigrade and may release specimens for purposes of confidential, anonymous scientific study unless prohibited by the parents, legal guardians, or children from whom the specimens were obtained when the children are eighteen years of age or older.

4. Hospital staff or other persons who collect these specimens shall ensure that the parent's or legal guardian's storage choice is documented on the Blood Sample Storage Options form if the parent or legal guardian does not agree to have their child's blood specimen stored and potentially released for confidential, anonymous scientific study. In these instances, the Laboratory shall maintain all such specimens based upon the storage option chosen by the parent or legal guardian as documented on the Blood Sample Storage Options form.

**Section G - Use of Stored Specimen**

1. Stored blood specimens may be released for the purposes of confidential, anonymous scientific study unless prohibited by the parent, legal guardian, or child from whom the specimen was obtained when he/she is eighteen years of age or older.

2. The Department's Institutional Review Board shall approve all scientific studies that use stored blood specimens before the specimens are released.

3. Blood specimens released for scientific study shall not contain information that may be used to determine the identity of the children from whom they were obtained by the person(s) to whom the specimens are released. The Department shall code the specimens before releasing them so that the Department can identify the children from whom the blood specimens were obtained if necessary.

4. If any such scientific study identifies genetic or other information that may benefit the children from whom the specimens were obtained, the Department may confidentially provide this information to the parents, legal guardians or children from whom the specimens were obtained when the children are eighteen years of age or older.

**Section H - Forms**

1. **Religious Objection Form**: The Religious Objection Form, Appendix A of this regulation, shall be completed if the parents refuse newborn screening for inborn metabolic errors and hemoglobinopathies for their child based upon religious convictions.

2. **Information Release Form**: The Information Release Form, Appendix B of this regulation, may be completed as needed for release of information regarding newborn screening for inborn metabolic errors and hemoglobinopathies to persons other than those specified elsewhere in this regulation.

3. **Blood Sample Storage Options Form**: The Blood Sample Storage Options Form, Appendix C of this regulation, shall be completed if the parents or legal guardians do not agree to have their child's specimen stored and potentially released for confidential, anonymous scientific study.

**Section I - Enforcement Provision**

1. **Constitutionality**

If any part or provision of these regulations is legally declared unconstitutional or if the application thereof to any persons or circumstances is held invalid, the validity and constitutionality of the remainder of these regulations shall not be affected thereby.

2. **Penalties**

Violation of these regulations shall be punishable in accordance with Section 44-37-30 of the Code of Laws of South Carolina, 1976, as amended.

**APPENDIX A**: Religious Objection Form: DHEC 1804, Newborn Screening Program, Parental Statement of Religious Objection

I am the parent or legal guardian of _____, a child born _____ in South Carolina. I request that my child not be tested by blood spot screening in order to detect silent, deadly metabolic diseases and hemoglobinopathies. I certify that this refusal is based on religious grounds. Religious grounds are the only permitted reason for refusal under South Carolina law, Section 44-37-30 (C).

I understand that my child may suffer brain damage, other bodily harm or death if a disease that can be detected by blood spot screening is not diagnosed. I understand that such harm can be lessened or prevented by early diagnosis and treatment. I understand that these diseases are usually silent and may be present in a child that looks healthy.

I understand that the blood spot screening test is the best way to detect these disorders early, and that testing is routinely done for every child. I understand that this testing is quick, easy and that the results are confidential. I understand that this testing has been the standard of care for all children born in South Carolina and the rest of the United States for many years.

I have been fully informed of, and fully understand, the possible devastating consequences to my child's health if blood spot screening is not done. I have been fully informed of, and fully understand the benefits of testing and blood specimen storage. I have been given the brochure produced by the South Carolina Department of Health and Environmental Control that describes the conditions for which testing is currently available and explains the benefits of testing and blood specimen storage.

I also understand that my child would have been tested for these conditions except for my objection. I have been given the opportunity to ask questions concerning this testing and these conditions, and all of my questions have been fully answered to my satisfaction.

I release and hold harmless the South Carolina Department of Health and Environmental Control, the hospital or other facility at which the birth occurred, the person(s) responsible for the collection of the blood spots, and any other person or entity relying on this objection, for any injury, illness and/or consequences, including the death of my child, which may result to my child as the result of my refusal of blood spot screening.

Parent: _____ Date: _____

Witness: _____

NOTE TO PROVIDERS: This form is only necessary if the parent or legal guardian refuses testing for inborn metabolic errors and hemoglobinopathies.

**APPENDIX B**: Information Release Form: DHEC 1878, Authorization to Release Information Relative to Newborn Screening for Inborn Metabolic Errors and Hemoglobinopathies

Please check all boxes that apply.

- ❑ A. I agree that information about _____, born _____, obtained as a result of tests conducted for screening for inborn metabolic errors and hemoglobinopathies may be released or exchanged with the following providers:

  _____

  _____

  _____

- ❑ B. In cases where this information is immediately needed for continuity of health care, I authorize the South Carolina Department of Health and Environmental Control to provide this information to the providers listed above by fax.

- ❑ C. I authorize my signed form to be faxed to the providers listed above.

I understand that my confidentiality cannot be guaranteed when sending this information by fax. I understand that the copy of my signature below may be treated as an original signature.

I am the client, parent or legal guardian. I understand that I am responsible for this information if it is released to me and that my records are protected generally under state laws as well as statutes governing specific types of information and cannot be disclosed without my authorization. I also understand that I may revoke this authorization at any time except to the extent that action has been taken on it.

Signature: _____ Date: _____

Witness: _____ Date: _____

Revoked: _____Date: _____

*Some babies are born with diseases of the blood or body function. A baby with one of these diseases looks healthy. However, these diseases can cause mental retardation, abnormal growth, infections, or death. Some of these diseases can be found by early testing. This testing, called newborn screening, is important so that your baby is not harmed by one of these diseases. During newborn screening, a small sample of your baby's blood is taken from the heel. The blood is tested. The blood shows if your baby has any of the "newborn screening" diseases. If your baby has one of these diseases, your doctor can treat your baby.*

*DHEC can store your baby's blood sample for special study. Studies help DHEC find out new information about diseases. If a study finds something in your child's blood sample that can help your child, DHEC can confidentially notify you (or your child if he/she is 18 years or older).*

**APPENDIX C**: Blood Sample Storage Options Form: DHEC 1812, Blood Sample Storage Options, Screening for Inborn Metabolic Errors and Hemoglobinopathies

Child's complete legal name: _____

Child's date of birth: _____

Parent or legal guardian's complete name: _____

Parent or legal guardian's complete address: _____

South Carolina law requires the Department of Health and Environmental Control to store your child's blood sample in a manner required by law. The blood sample is collected on a special piece of filter paper. This is called "newborn screening." The blood is tested to see if your child has one of the "newborn screening" diseases that can cause mental retardation, abnormal growth or even death. After the tests are done, the filter paper is stored in a freezer at the state laboratory. This storage is highly protected, and each sample is held under strict confidentiality.

A child's blood sample can only be released for approved research, without any identifying information, to learn new information about diseases. The law allows you to choose one of the options below, if you do not want your child's blood sample handled this way. **However, you are not required to check one of the boxes below.**

- ❑ I want my child's blood sample stored by the South Carolina Department of Health and Environmental Control, but I do not want my child's blood sample to be used for research.

- ❑ I want my child's blood sample destroyed by the South Carolina Department of Health and Environmental Control two years after the date of testing.

- ❑ I want my child's blood sample to be returned to me two years after the date of testing. I understand that it is my responsibility to notify the South Carolina Department of Health and Environmental Control, 2600 Bull Street, Columbia, SC, 29201, of address or name changes.

I have been given the brochure produced by the South Carolina Department of Health and Environmental Control that describes the conditions for which testing is currently available and explains the benefits of testing and blood sample storage.

Parent: _____ Date: _____

I have given the brochure produced by the South Carolina Department of Health and Environmental Control to the parent/legal guardian of the child named above.

Name: _____ Date: _____

*DHEC can store your baby's blood sample for special study. Studies help DHEC find out new information about diseases. If a study finds something in your child's blood sample that can help your child, DHEC can confidentially notify you (or your child if he/she is 18 years or older).*

IF THIS FORM IS NOT SIGNED BY A PARENT/LEGAL GUARDIAN AND/OR NONE OF THE ABOVE BOXES ARE CHECKED, THE BLOOD SAMPLE WILL BE STORED AS REQUIRED BY SC CODE ANN. SECTION 44-37-30 AT –20 DEGREES CENTIGRADE AND MAY BE RELEASED ONLY FOR CONFIDENTIAL, ANONYMOUS SCIENTIFIC STUDY.

NOTE TO PROVIDERS: The parent or legal guardian is not required to sign this form. However, the person who gives the brochure that explains neonatal testing and blood sample storage to the parent or legal guardian must sign this form

# DHEC 1804, Parental Statement of Religious Objection Instructions

PURPOSE: This form is used by hospital, health department and other health care provider staffs to document a religious objection to newborn screening for inborn errors of metabolism and hemoglobinopathies.

ITEM BY ITEM INSTRUCTIONS:

Top Section: Print parents or guardians' names on the line indicated. Print child's name and date of birth on the lines indicated.

Bottom Section: The parent or guardian signs his/her name and indicates the date in the appropriate space. The witness signs his/her name and indicates the date in the appropriate space.

OFFICE MECHANICS AND FILING: Mail the original to: Newborn Screening Program, Division of Children's Health and Perinatal Services, SC DHEC, Mills/Jarrett Complex, Box 101106, Columbia, SC 29211.

One copy can be given to the parent or guardian. One copy is filed under consents at the health department/facility where the form was signed. The form should be retained according to the medical records retention schedule.

D H E C

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## Newborn Screening Program
## Parental Statement of Religious Objection

I am the parent or legal guardian of _____, a child born _____ in South Carolina. I request that my child not be tested by blood spot screening in order to detect silent, deadly metabolic diseases and hemoglobinopathies. I certify that this refusal is based on religious grounds. Religious grounds are the only permitted reason for refusal under South Carolina law,
Section 44-37-30(C).

I understand that my child may suffer brain damage, other bodily harm or death if a disease that can be detected by blood spot screening is not diagnosed. I understand that such harm can be lessened or prevented by early diagnosis and treatment. I understand that these diseases are usually silent, and may be present in a child that looks healthy. I understand that the blood spot screening test is the best way to detect these disorders early, and that testing is routinely done for every child. I understand that this testing is quick, easy and that the results are confidential. I understand that this testing has been the standard of care for all children born in South Carolina and the rest of the United States for many years.

I have been fully informed of, and fully understand, the possible devastating consequences to my child's health if blood spot screening is not done. I have been fully informed of, and fully understand the benefits of testing and blood specimen storage. I have been given the brochure produced by the South Carolina Department of Health and Environmental Control that describes the conditions for which testing is currently available and explains the benefits of testing and blood specimen storage. I also understand that my child would have been tested for these conditions except for my objection. I have been given the opportunity to ask questions concerning this testing and these conditions, and all of my questions have been fully answered to my satisfaction.

I release and hold harmless the South Carolina Department of Health and Environmental Control, the facility at which the birth occurred, the person(s) responsible for the collection of the blood spots, and any other person or entity relying on this objection, for any injury, illness and/or consequences, including the death of my child, which may result to my child as the result of my refusal of blood spot screening.

Parent: _____ Date: _____

Witness: _____

NOTE TO PROVIDERS: This form is only necessary if the parent or legal guardian refuses testing for inborn metabolic errors and hemoglobinopathies.

# Other Resources

The **Advisory Committee on Heritable Disorders in Newborns and Children** (**ACHDNC**) was established under the Public Health Service Act, Title XI, § 1109 (42 U.S.C. 300b-10), as amended by the Newborn Screening Saves Lives Reauthorization Act of 2014 (P.L. 113-240).

The Committee advises the Secretary, U.S. Department of Health and Human Services (HHS) on the most appropriate application of universal newborn screening tests, technologies, policies, guidelines, and standards.

https://www.hrsa.gov/advisory-committees/heritable-disorders/index.html

**American College of Medical Genetics and Genomics (ACMG)** ACT sheets provide immediate steps for physicians to take upon receiving a positive screen for an infant in his or her practice. https://www.acmg.net

**Baby's First Test** provides current educational and family support and services information, materials, and resources about newborn screening at the local, state, and national levels and serves as the Clearinghouse for newborn screening information. https://www.babysfirsttest.org

**The Centers for Disease Control (CDC) and Prevention,** Newborn Screening Portal serves as the national resource for newborn screening activities and data to prevent death or disability and enable children to reach their full potential. https://www.cdc.gov/newbornscreening

**The Clinical and Laboratory Standards Institute (CLSI)** provides resources on specimen collection and newborn screening. https://clsi.org

The mission of the **Cystic Fibrosis Foundation** is to cure cystic fibrosis and to provide all people with the disease the opportunity to lead full, productive lives. https://www.cff.org

**The Immune Deficiency Foundation (IDF),** is a national nonprofit patient organization dedicated to improving the diagnosis, treatment and quality of life of persons with Primary immunodeficiency diseases (PI) such as Severe Combined Immune Deficiency (SCID) and others, through advocacy, education and research. https://primaryimmune.org

**The March of Dimes** leads the fight for the health of all mothers and babies and has helped millions of babies survive and thrive.

https://www.marchofdimes.org/baby/newborn-screening-tests-for-your-baby.aspx

**National Organization for Rare Disorders (NORD)** is a patient advocacy organization dedicated to individuals with rare diseases and the organizations that serve them. https://rarediseases.org

# Hearing Loss (HL) and Critical Congenital Heart Defects (CCHD)*

*These point of care newborn screening tests (not blood tests) are administered at the hospital or other birthing facility.

For newborn hearing screening and hearing loss information, please refer to SC DHEC First Sound Hearing Screening Program. For CCHD information refer to the SC DHEC Birth Defects Program.

First Sound Program Manager/Audiologist:
Tara Carroll, MCD, CCC/A.....803-898-0708
email: carroltp@dhec.sc.gov

Birth Defects Program Manager:
Vinita Oberoi Leedom, MPH, APM, PMP.....803-898-0771
email: leedomvo@dhec.sc.gov

## Acknowledgements

The metabolic disorders information pages were adapted from the following sources:

ACMG ACT Sheets and Confirmatory Algorithms

Fact Sheets, Oregon State Public Health Laboratory

Health Professionals Guide to Newborn Screening, Wisconsin Newborn Screening Laboratory

New England Consortium of Metabolic Programs at Children's Hospital Boston